

Assuring Quality in Newborn Screening Systems

Marrakech – November 14, 2006

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Newborn Screening is
MORE than a blood test!



Newborn Screening Is:

A System that includes

- Medical Practitioners (Doctors, Nurses, Mid-wives)
- Laboratory Personnel
- Administrative and Follow-up Personnel
- Specialty Care Providers
- Source(s) of Payment
- Family Members
- Other Interested People (e.g. Policy Makers)



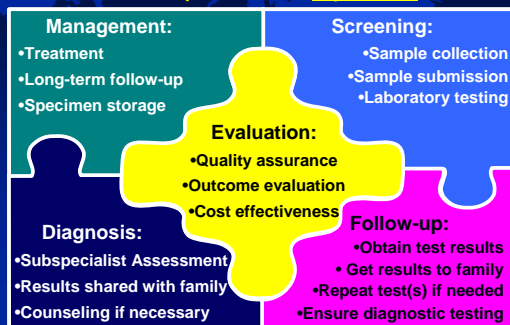
Newborn Screening Is:

Organized Differently in Different Countries

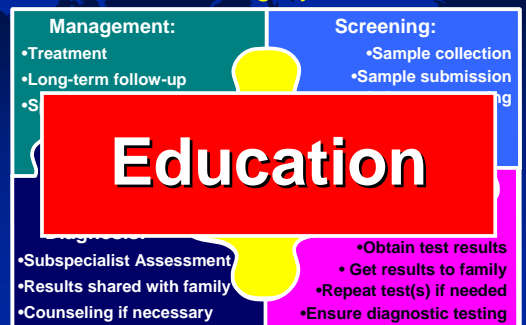
- Usually begins as a pilot for limited conditions on a local level – state, city, etc.
- Often results in a national policy of the Health Ministry.
- Sometimes implementation is local.
- May be required by law or proclamation (usually with signed dissent).
- May be required to be offered by person responsible for birthing process (consent).



Newborn Screening Must Be Developed As A System



Components of the Newborn Screening System



National Newborn Screening Information System (NNSIS)

National Newborn Screening Information System

Home | NNSGRC Home | Help on NNSIS | NNSGRC Staff Only | Contact NNSGRC | Logout

Welcome to the National Newborn Screening Information System (NNSIS) database.

The database is hosted by the **National Newborn Screening and Genetics Resource Center (NNSGRC)** and is designed to provide a secure, Internet based, real-time, information collection and reporting system for capturing state and territorial newborn screening information.

The system uses existing reporting requirements specified in the former National Newborn Screening Annual Report.

- Obtain a Report
- Enter Individual or Annual Case Information
- Contact NNSGRC

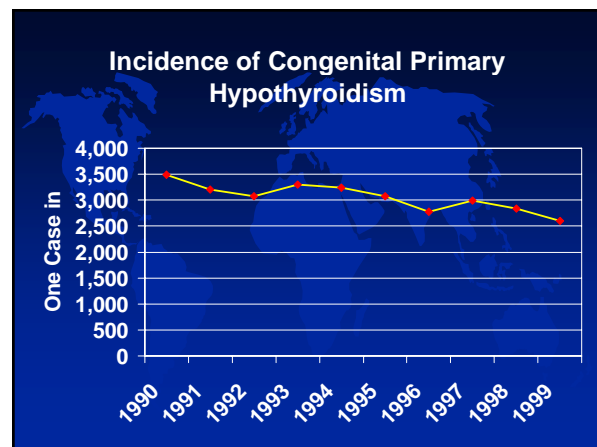
Welcome to our Newborn Screening Data System

U.S. Newborn Screening Data 1990-1999

DISORDER	SCREENED	CASES	INCIDENCE
S,S + SC Disease	28,149,621	11,376	1:2,474
Primary Hypothyroidism	40,214,946	13,213	1:3,044
S,S Disease	28,149,621	7,565	1:3,721
Cystic Fibrosis	1,459,834	372	1:3,924
S,C Disease	28,149,621	3,811	1:7,386
Toxoplasmosis	989,402	95	1:10,415
Classical PKU	40,028,546	2,098	1:19,079

U.S. Newborn Screening Data 1990-1999

DISORDER	SCREENED	CASES	INCIDENCE
Classical PKU	40,028,546	2,098	1:19,079
PKU Variant Clin. Sig.	40,028,546	772	1:51,850
Clin. Sig. Hyperphe	40,028,546	2,870	1:13,947
Classical CAH	13,347,888	703	1:18,987
Classical Galactosemia	35,897,634	674	1:53,261
Biotinidase	12,754,403	208	1:61,319
MSUD	13,801,657	60	1:230,028
Homocystinuria	12,027,751	35	1:343,650



Newborn Screening Is:

A System that demands Quality and Quality Assurance

- Educational materials and other program information
- Birthing facility practices
- Specimen transport systems
- Record keeping and record keeping systems
- Laboratory processes
- Result transmittal and follow-up actions
- Medical and other program services

Are parent materials accurate? readable? distributed in a timely way? useful?

Are professional materials accurate? readable? distributed in a timely way? useful?

PRIMARY TSH TEST/ ELEVATED TSH
Congenital Hypothyroidism

Newborn Screening ACT Sheet
[Primary TSH test/Elevated TSH]
Congenital Hypothyroidism

Differential Diagnosis: Primary congenital hypothyroidism (CH), transient CH.

Clinical Description: Lack of adequate thyroid hormone production.

You Should Take the Following Actions:

- Contact family **IMMEDIATELY** to inform them of the newborn screening test result.
- Consult pediatric endocrinologist, referral to endocrinologist if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

Diagnostic Evaluation: Diagnostic tests should include serum free T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Test results include reduced free T4 and elevated TSH in primary hypothyroidism; if done, reduced total T4 and low or normal T3 resin uptake.

Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy face, large fontanelles, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

Additional Information:
Click on the name to take you to the website. Complete URLs are listed in the Appendix.
[New England Newborn Screening Program](#)
[American Academy of Pediatrics](#)
[Genetics Home Reference](#)

Referred (local, state, regional and nationally):
[Larissa M. White, Pediatric Endocrinologist, "Find a Doc"](#)
Contact local/regional University-affiliated medical center

In 2004, HRSA funded development of a self-assessment tool for use in evaluating the quality of the elements of a comprehensive newborn screening system.

Performance Evaluation and Assessment Scheme

For Newborn Screening

<http://genes-r-us.uthscsa.edu>

NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

SELF-ASSESSMENT PERFORMANCE CHECKLIST
With Example References/Activities for Improvement

I. CROSS-CUTTING
II. PRE-ANALYTICAL
III. ANALYTICAL
IV. POST-ANALYTICAL
REFERENCES

PERFORMANCE INDICATOR	FINDINGS			EXAMPLES/REFERENCES
	Yes	In Prep	No	
	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

PEAS Performance Indicators

http://genes-r-us.uthscsa.edu/PEAS.doc

4. **PEAS Performance Indicators and Assessment** - In order for the newborn screening system to function, there are critical administrative and financial issues that must be addressed and an effective system for ongoing monitoring, auditing, and improvement of performance is necessary. Planning and administrative considerations must be completed and addressed in order to ensure the system is ready for future growth.

Performance Indicator	Findings		Comments
	Yes	In Progress	
1. Administration			
2. Laboratory			
3. Information Management			
4. Quality Improvement			
5. Financial Management			
6. Regulatory Compliance			
7. Public Relations			
8. Evaluation and Assessment			

NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

I. GENERAL NEWBORN SCREENING SYSTEM CONSIDERATIONS

A. Education

1. Education Plan

NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

I. GENERAL NEWBORN SCREENING SYSTEM CONSIDERATIONS

A. Education

1. Education Plan

B. Computerized Information System

1. Scope

2. Integrity

NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

I. GENERAL NEWBORN SCREENING SYSTEM CONSIDERATIONS

- A. Education
 - 1. Education Plan
- B. Computerized Information System
 - 1. Scope
 - 2. Integrity
- C. Monitoring of Timely and Universal Screening
 - 1. Required Screening
 - 2. Subsequent Screens



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

I. GENERAL NEWBORN SCREENING SYSTEM CONSIDERATIONS (continued)

- D. Program Administration and Financing
 - 1. Administration
 - 2. Financing



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

I. GENERAL NEWBORN SCREENING SYSTEM CONSIDERATIONS (continued)

- D. Program Administration and Financing
 - 1. Administration
 - 2. Financing
- E. Contingency Plan
 - 1. Program Administration
 - 2. Laboratory
 - 3. Follow-up



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

II. PRE-ANALYTICAL CONSIDERATIONS

- A. Personnel
 - 1. Personnel Training, Recruitment, and Retention
 - 2. Personnel Competency



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

II. PRE-ANALYTICAL CONSIDERATIONS

- A. Personnel
 - 1. Personnel Training, Recruitment, and Retention
 - 2. Personnel Competency
- B. Prenatal Education
 - 1. Preparation/Distribution of PARENT Education Materials
 - 2. Preparation/Distribution of PROFESSIONAL Education Materials



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

II. PRE-ANALYTICAL CONSIDERATIONS (continued)

- C. Screening Process
 - 1. Specimen Collection Device
 - 2. Specimen Collection/Transmittal
 - 3. Specimen Receipt
 - 4. Specimen Tracking



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

II. PRE-ANALYTICAL CONSIDERATIONS (continued)

- C. Screening Process
 - 1. Specimen Collection Device
 - 2. Specimen Collection/Transmittal
 - 3. Specimen Receipt
 - 4. Specimen Tracking
- D. Laboratory Safety
 - 1. Employee Training
 - 2. Safety Program



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

III. ANALYTICAL CONSIDERATIONS

- A. Testing Process
 - 1. Procedures Manual
 - 2. Quality Assurance Program



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

III. ANALYTICAL CONSIDERATIONS

- A. Testing Process
 - 1. Procedures Manual
 - 2. Quality Assurance Program
- B. Laboratory Instrumentation
 - 1. Instrument Operations
 - 2. Quality Assurance



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

III. ANALYTICAL CONSIDERATIONS

- A. Testing Process
 - 1. Procedures Manual
 - 2. Quality Assurance Program
- B. Laboratory Instrumentation
 - 1. Instrument Operations
 - 2. Quality Assurance
- C. Laboratory Supplies/Reagents
 - 1. Quantity
 - 2. Quality



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

III. ANALYTICAL CONSIDERATIONS

- A. Testing Process
 - 1. Procedures Manual
 - 2. Quality Assurance Program
- B. Laboratory Instrumentation
 - 1. Instrument Operations
 - 2. Quality Assurance
- C. Laboratory Supplies/Reagents
 - 1. Quantity
 - 2. Quality
- D. Working Environment
 - 1. Working Environment



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

IV. POST-ANALYTICAL CONSIDERATIONS

- A. Screening Test Results
 - 1. Laboratory Assay Documentation
 - 2. Laboratory Result Reporting
 - 3. Records Storage (including reports, results, residual specimens)



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

IV. POST-ANALYTICAL CONSIDERATIONS

- A. Screening Test Results
 1. Laboratory Assay Documentation
 2. Laboratory Result Reporting
 3. Records Storage (including reports, results, residual specimens)
- B. Short-Term Follow-up
 1. Written Follow-up Procedures
 2. Follow-up Communications



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

IV. POST-ANALYTICAL CONSIDERATIONS (continued)

- C. Follow-up Support Activities
 1. Diagnosis
 2. Parent Education
 3. Counseling (Including Nutritional, Genetic etc.)
 4. Medical Management



NEWBORN SCREENING PERFORMANCE EVALUATION AND ASSESSMENT

IV. POST-ANALYTICAL CONSIDERATIONS (continued)

- C. Follow-up Support Activities
 1. Diagnosis
 2. Parent Education
 3. Counseling (Including Nutritional, Genetic etc.)
 4. Medical Management
- D. Program Evaluation
 1. Short-Term Program Evaluation
 2. Long-Term Program Evaluation (Including Medical Management)



Building Individual Country (Program) PEAS

Developing PEAS in the Philippines



Philippine PEAS

- ◆ Indicators for the Newborn Screening Centers (laboratories)
- ◆ Indicators for the Center for Health Development (Regional Implementers)
 - Evaluation of NBS Program implementation in the Region
- ◆ Indicators for the Newborn Screening Health Facilities (hospitals and community health centers)
 - Assessment of the NBS Program in the Health Facilities

Program Components Reviewed

1. Membership
2. Purchase Orders
3. Handling and Transport
4. Processing
5. Billing and Collection
6. Inventory
7. Releasing of Results
8. Generation of Reports
9. Program and Laboratory Administration

